

GENOMICS: WHY SHOULD WE CARE?

The importance of being: genomic literate
genomic informed



Health Literacy

Orphan Black



Orphan Black video

News Headlines

Scientists Say They Hope To Create A
Human Genome In The Lab

Humans will be 'irrevocably altered' by genetic editing, warn scientists ahead of summit

An open letter from 150 scientists, campaigners and health experts is calling for a worldwide ban on genetic editing ahead of a summit in Washington

**The Genetic Tool That Will
Modify Humanity**

Crispr allows scientists to control the blueprints of life, for better or worse.

British Scientists Seek Permission To Edit
DNA In Human Embryos

**Clinical Genetics Has a Big
Problem That's Affecting
People's Lives**

Unreliable research can lead families to make health decisions they might regret.

Genetically Modified Humans?
How Genome Editing Works

Having a baby? Best to run a gene screening test

Health professional literacy

- Most physicians have no formal training in genetics
- Little research has focused on their understanding of the recent developments in genomics
- Patients ill-informed, inflated expectations, little scientific evidence regarding clinical utility of genomic interventions
- Health professionals have not always kept up to date with the genetic advances
- Need to improve medical education and beyond initial training

Leading causes of death

1. Heart disease: 614,348
2. Cancer: 591,699
3. Chronic lower respiratory diseases: 147,101
4. Accidents (unintentional injuries): 136,053
5. Stroke (cerebrovascular diseases): 133,103
6. Alzheimer's disease: 93,541
7. Diabetes: 76,488
8. Influenza and pneumonia: 55,227
9. Nephritis, nephrotic syndrome, and nephrosis: 48,146
10. Intentional self-harm (suicide): 42,773

Definitions

□ Genomic Health Literacy

- ▣ The capacity to obtain, process, understand, and use genomic information for health related decision making.

□ Genomic Science Literacy

- ▣ The knowledge of basic genetics and genomics concepts and processes needed to build conceptual understanding, and the necessary mathematical knowledge to support this comprehension.

Genomic literacy



- Genomic health literacy
- Genomic science literacy
- Role of media in genomic literacy

Genomic Health Literacy

- ❑ Lack biology basics
- ❑ Lack mathematical concepts
- ❑ Low health literacy



Genomic Science Literacy

- ❑ K-12 education unable to keep up with scientific advancements
- ❑ Low emphasis on genomics
- ❑ Some teachers have misconceptions about genetics/genomics and little understanding
- ❑ Teachers need updated skills and have little access to genetic/genomic quality science curriculum
- ❑ Encourage partnerships with scientists

Media role in genomic literacy

- ❑ Scientists lack training when communicating with media and general public
- ❑ Popular and mass media lack knowledge and often relay incorrect information
- ❑ Educators and researchers need to adapt and learn to inform through newer media platforms such as social media and podcasts



HHS Public Access

Author manuscript

Genet Med. Author manuscript; available in PMC 2014 July 30.

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What Does it Mean to be Genomically Literate? National Human Genome Research Institute Meeting Report

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[PMID 23448722](https://pubmed.ncbi.nlm.nih.gov/23448722/)

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4115323/>

Genomic discoveries will increasingly advance the science of medicine. Limited genomic literacy may adversely impact the public's understanding and use of the power of genetics and genomics in health care and public health. In November 2011, a meeting was held by the National Human Genome Research Institute to examine the challenge of achieving genomic literacy for the general public, from K-12 to adult education. The role of the media in disseminating scientific messages and in perpetuating, or reducing, misconceptions was also discussed. Workshop participants agreed that genomic literacy will only be achieved through active engagement between genomics experts and the varied constituencies that comprise the public.

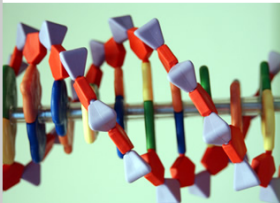
NIH National Human Genome Research Institute


NIH National Human Genome Research Institute
Advancing human health through genomics research


SEARCH GENOME.GOV


Research Funding | Research at NHGRI | Health | **Education** | Issues in Genetics | Newsroom | Careers & Training | About | Español


Education Educational materials about genetics and genomics


 **GENOME UNLOCKING** Smithsonian NHGRI Genome Exhibition


 All About The Human Genome Project


 Talking Glossary of Genetic Terms
Genetic terms, illustrations


 Genomic Careers
Find out about careers in genomics and genetics

 National DNA Day
April 25, 2016

 Genetic Education Resources for Teachers


 Online Genetic Education Resources
Links to other sites

 Fact Sheets
DNA, cloning, genomics and more


 Education Resources
Online Education Kit: Understanding the Human Genome Project



Feature
Video now available
Harry Potter and the Genetics of Wizarding

 For NHGRI's National DNA Day inaugural speaker series, Eric P. Spana, Ph.D. from Duke University presented Harry Potter and the

Highlights
National DNA Day: Improving genomic literacy on a national scale

 The National Human Genome Research Institute marked its fourteenth annual National DNA Day by organizing a nationwide network of educational events for students, teachers and


See Also:
Education
GenomeTV
Genome
Education
On Other Sites

Health Information about genetics and genomics, rare diseases, patient care and more


For Patients and the Public
Community Engagement and Community Health
Family History
Genetics & Genomics Science & Research
Genetic & Rare Diseases Information Center
Genomic Medicine and Health Care
Online Health and Support Resources
Specific Genetic Disorders

For Health Professionals
Competency & Curricular Resources
Genetics 101
Inter-Society Coordinating Committee (ISCC)
New Horizons and Research
Patient Management
Policy and Ethics Issues
What is Genomic Medicine?

Feature
Apply now for the "short course" health professionals track

 The Genomic Healthcare Branch is offering the NHGRI Short Course in Genomics: Nurse, Physician Assistant and Faculty Track from August 1 - August 3, 2016. This year's course is for nurses, nurse practitioners, physician assistants and the faculty who educate these health professionals in genomics. [Read more](#)

Highlights
Video: A G2C2 Website Overview

 This introduction to the Genetics/Genomics Competency Center (G2C2) website at <http://g-2-c-2.org/>, gives new users an opportunity to view key features of this centralized collection of genomics educational resources for healthcare educators and providers. [Read more](#)

Genomic knowledge is power in the fight against obesity

Webinars for Health Insurers and Payers: Understanding Genetic Testing

See Also
[GenomeTV](#)
[Genomic Healthcare Branch](#)
[Fact Sheets](#)
[Genetic Education Resources for Teachers](#)
[All About the Human Genome Project](#)
[Health Archive](#)

On Other Sites:
[YouTube](#) [GenomeTV](#)
NHGRI's YouTube channel

G2C2 <http://g-2-c-2.org//>

National Coalition for Health Professional Education in Genetics (NCHPEG)

The screenshot shows the NCHPEG website homepage. At the top is a red navigation bar with links: HOME, ABOUT NCHPEG, PRODUCTS AND PROGRAMS, ANNUAL MEETING, and NCHPEG EVENTS. Below the navigation bar are the NCHPEG logo (a stylized DNA helix) and The Jackson Laboratory logo (a stylized 'J' with a person icon). A search bar is located to the right of the logos. The main content area features a large graphic with the text 'HEALTH 2.0' in a cloud, surrounded by phrases like 'SHARING DATA FOR RESEARCH', 'COLLABORATION AND PRACTICE', 'STAYING INFORMED', 'MANAGING A PARTICULAR DISEASE', and 'MEDICAL EDUCATION'. To the right of this graphic is a sidebar with three sections: 'Collaborate' (with a 'Comment' icon), 'Educate' (with a 'Comment' icon), and 'Evaluate' (with a 'Good' icon). Below the main graphic is a 'PROGRAM HIGHLIGHTS' section with three items: 'Medicine's Future: Genomics for Practicing Providers', 'GeneFacts', and 'Colorectal Cancer: Is Your Patient at High Risk?'. To the right of this section is a 'Pregnancy & Health Profile' section with a tree diagram and a 'DOWNLOAD HERE' button.

HOME ABOUT NCHPEG PRODUCTS AND PROGRAMS ANNUAL MEETING NCHPEG EVENTS

NCHPEG
National Coalition
for Health Professional
Education in Genetics

**The Jackson
Laboratory**
Leading the search
for tomorrow's cures

SHARING DATA
FOR RESEARCH

COLLABORATION
AND PRACTICE

STAYING
INFORMED

HEALTH 2.0

MANAGING
A PARTICULAR
DISEASE

Medicine's Future: A
Genomics Curriculum for
Clinicians

Engaging clinicians in continuing
education to integrate genomics
into th...

Read more

GeneFacts

GeneFacts is a point-of-care,
decision-support system for non-
geneticists...

Read more

Colorectal Cancer: Is Your
Patient at High Risk?

Learn to identify patients at
increased risk for hereditary
colorectal c...

Read more

Pregnancy & Health Profile

Genetic Screening, Risk Assessment
and Clinical Decision Support

DOWNLOAD
HERE

NLM GeneEd

The screenshot shows the NLM GeneEd website homepage. At the top, there is a yellow banner with the GeneEd logo (a stylized DNA double helix) and the tagline "Genetics, Education, Discovery". To the right of the logo, there are links for "Text Size +", "Print This Page", and "Email Us". Below the banner is a navigation bar with buttons for "Home", "Topics", "Labs & Experiments", "Teacher Resources", "Careers in Genetics", and "Highlights". A search bar is located on the right side of the navigation bar. The main content area is divided into a grid of topic boxes, each with a small image and a title. The topics include: Cell Biology (The study of the cell, including mitosis and meiosis), Genetic Conditions (Conditions caused by gene variations or mutations), DNA Forensics (The application of genetic testing for legal purposes), DNA, Genes, Chromosomes (The building blocks of inheritance), Evolution (Processes by which organisms are changed over time), Top Issues In Genetics (Select genetic topics of popular interest), Heredity/Inheritance Patterns (The genetic transmission of traits), Biostatistics (The use of mathematics to investigate life science related problems), Epigenetics/Inheritance and the Environment (The genome's dynamic response to the environment), and Biotechnology (Biological techniques used to enhance products). On the right side of the main content area, there is a large image of two young women looking at a laptop, with a DNA double helix graphic overlaid.

GeneEd *Genetics, Education, Discovery*

Text Size +
Print This Page
Email Us

Home Topics Labs & Experiments Teacher Resources Careers in Genetics Highlights search... GO

Cell Biology
The study of the cell, including mitosis and meiosis

Genetic Conditions
Conditions caused by gene variations or mutations

DNA Forensics
The application of genetic testing for legal purposes

DNA, Genes, Chromosomes
The building blocks of inheritance

Evolution
Processes by which organisms are changed over time

Top Issues In Genetics
Select genetic topics of popular interest

Heredity/Inheritance Patterns
The genetic transmission of traits

Biostatistics
The use of mathematics to investigate life science related problems

Epigenetics/Inheritance and the Environment
The genome's dynamic response to the environment

Biotechnology
Biological techniques used to enhance products

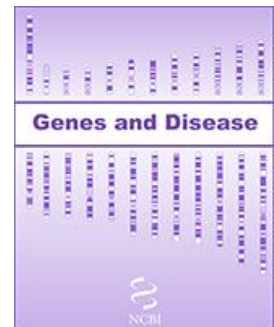
NCBI Bookshelf

Genes and Diseases

- ❑ Organized by the parts of the body that genetic disorders affect
- ❑ Over 80 genetic disorder summaries
- ❑ Images and interesting facts
- ❑ PDF downloads of chapters
- ❑ Links to related research literature and pertinent websites

[Genes and Diseases](http://www.ncbi.nlm.nih.gov/books/NBK22183/)

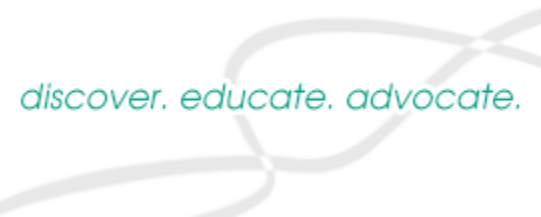
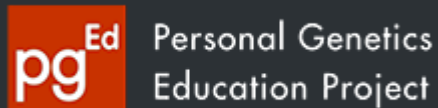
<http://www.ncbi.nlm.nih.gov/books/NBK22183/>



Literacy/Education Resources



Public Health Genomics



Tips for communicating to patients

- Listen, pay attention, respond
- Use plain language
- Use patient's words
- Slow down
- Limit and repeat content
- Show examples
- Invited patient participating
- Use teach back

[Health Literacy and Precision Medicine](http://www.nationalacademies.org/hmd/Activities/PublicHealth/HealthLiteracy/2016-MAR-2.aspx)

[http://www.nationalacademies.org/hmd/Activities/
PublicHealth/HealthLiteracy/2016-MAR-2.aspx](http://www.nationalacademies.org/hmd/Activities/PublicHealth/HealthLiteracy/2016-MAR-2.aspx)

Tips for communicating to healthcare professionals

- Bring a family member or a friend
- Write down or record the information
- Speak your mind
- Repeat the information as you understand it
- Ask questions (who, what, where, why, how)
- Ask for more information (website, printed handout, a library)

MedlinePlus Magazine

My Family Health Portrait

My Family Health Portrait, A tool from the Surgeon General

[Past Issues](#) / [Winter 2010 Table of Contents](#)



My Family Health Portrait is an Internet-based tool that makes it easy to create your family's health history. It is simple to fill out. It is private. It is valuable health information that you can share with family members, for their benefit, and with your healthcare practitioner, for your better health.

My Family Health History is available at:
<https://familyhistory.hhs.gov/fhh-web/home.action>

Using **My Family Health Portrait**, you can:

- Record your family's health history
- Print out and share the history with your family and your healthcare provider
- Save and regularly update your family health history for future use

Why is it important to know my family medical history?

Your family medical history is a record of health information about you and three generations of close relatives. Family history can be an important risk factor for problems like heart disease, stroke, diabetes, and cancer. A risk factor is anything that increases your chance of getting a disease. The reason a family history can help predict risk is that families share their genes, as well as other factors that affect health, like environment, lifestyles, and habits. A family medical history allows you to take steps to reduce your risk.



To Find Out More

MedlinePlus: Family History
www.nlm.nih.gov/medlineplus/familyhistory.html

Family Health History
www.cdc.gov/genomics/famhistory/index.htm

[MedlinePlus Magazine](#)

<https://www.nlm.nih.gov/medlineplus/magazine/issues/winter10/articles/winter10pg4.html>

My Family Health Portrait

 **National Human Genome Research Institute**
Advancing human health through genomics research

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[Home](#) > [Health](#) > [Genetics and Genomics for Patients and the Public](#) > **Family History: My Family Health Portrait**

Genetics and Genomics for Patients and the Public

- Community Engagement and Community Health Resources
- FAQ About Genetic Counseling
- FAQ About Genetic Disorders
- FAQ About Genetic Testing
- FAQ About Genetics, Disease Prevention and Treatment
- FAQ About Neglected Diseases
- FAQ About Pharmacogenomics
- FAQ About Rare Diseases
- Family History: My Family Health Portrait**

Family History



Find out how learning your family's health history can help you discover your genetic heritage and risks, and guide you in making healthy environment and lifestyle choices. Learn about how to obtain and create a family health history.

- [My Family Health Portrait](#)
- [Family History: Resources and Tools](#) [cdc.gov]
- [Family Health History Tools](#) [geneticalliance.org]
- [NIH SeniorHealth: Creating a Family Health History](#) [nihseniorhealth.gov]




NHGRI My Family Health Portrait <https://www.genome.gov/27527640/family-history-my-family-health-portrait/>

My Family Health History

My Family Health History








Update My Family History

On this screen you can:

- Use the tools to view your family tree diagram, find out your risks for certain diseases, or export your tree to help your close family member get started.
- Change your Family Health History by adding, removing or changing your relatives
 - Add information for a family member by pressing **Add History**  next to the name in the list.
 - Change your information or a family member's information by pressing **Update History**  next to the name in the list.
 - Remove a family member from your history by pressing **Remove**  next to the name in the list. (You cannot remove yourself, your parents, or grandparents.)

To find out more about what you can do, click on the 'Get Help' link on the menu bar above.

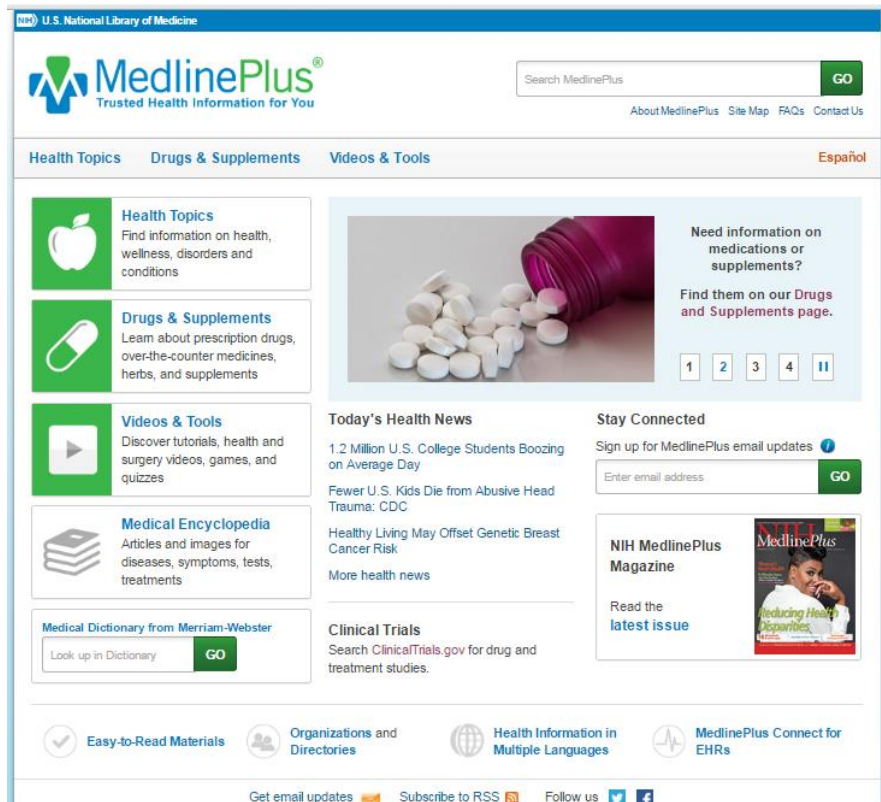
[Delete all Data and Restart](#) [Save Family History for Later Re-use](#) [Add Another Family Member](#) [View Diagram and Table](#)

Name	Relationship to me:	Add History	Update History	Remove Relative
My Family				
undefined	Self			
	Father			
	Mother			
My Father's Side of the Family				
	Paternal Grandfather			
	Paternal Grandmother			
My Mother's Side of the Family				
	Maternal Grandfather			
	Maternal Grandmother			
Recently Added Family Members				




Consumer Websites

MedlinePlus



- Health Topic pages:
 - Genetics
 - Genetic testing
 - Genetic counseling
 - Genetic disorders
 - Genes and therapy
- text word search

MedlinePlus



U.S. National Library of Medicine

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Health Topics Drugs & Supplements Videos & Tools Español

Home → Health Topics → Genetic Disorders

Genetic Disorders

On this page

Basics

- Summary
- Start Here
- Diagnosis and Tests
- Treatments and Therapies

Learn More

- Living With
- Related Issues
- Specifics
- Genetics

See, Play and Learn

- No links available

Research

- Statistics and Research
- Clinical Trials
- Journal Articles

Resources

- Reference Desk
- Find an Expert

For You

- Children
- Teenagers
- Women
- Patient Handouts



#ADAM

Get Genetic Disorders updates by email

Enter email address

GO

MEDICAL ENCYCLOPEDIA

[Alström syndrome](#)

[Basal cell nevus syndrome](#)

[Beriberi](#)

[Cystinuria](#)

[Genetics](#)

[Hemochromatosis](#)

Summary

Genes are the building blocks of heredity. They are passed from parent to child. They hold DNA, the instructions for making proteins. Proteins do most of the work in cells. They move molecules from one place to another, build structures, break down toxins, and do many other maintenance jobs.

Sometimes there is a mutation, a change in a gene or genes. The mutation changes the gene's instructions for making a protein, so the protein does not work properly or is missing entirely. This can cause a medical condition called a genetic disorder.

You can inherit a gene mutation from one or both parents. A mutation can also happen during your lifetime.

There are three types of genetic disorders:

Single gene disorders, when a mutation affects one gene. Single gene disorders are passed

Diagnosis and Tests

- How Are Genetic Conditions Diagnosed? [NH](#) (National Library of Medicine)
- MedlinePlus: Newborn Screening [NH](#) (National Library of Medicine)
Available in Spanish
- Progeria Research Foundation Diagnostic Testing Program (Progeria Research Foundation, Inc.)

Treatments and Therapies

- How Are Genetic Conditions Treated or Managed? [NH](#) (National Library of Medicine)

Living With

- Physical Therapy and Occupational Therapy in Progeria (Progeria Research Foundation, Inc.) - PDF

Related Issues

- Learning about an Undiagnosed Condition in an Adult [NH](#) (National Human Genome Research Institute)

Specifics

- Bloom's Syndrome (Chicago Center for Jewish Genetic Disorders)
- Chromosomal Conditions (March of Dimes Birth Defects Foundation)
Available in Spanish
- Cryopyrin-Associated Autoinflammatory Syndromes (CAPS) - Juvenile (American College of Rheumatology)
- Genetics Home Reference [NH](#) (National Library of Medicine)
- Learning about Poland Anomaly [NH](#) (National Human Genome Research Institute)
- Learning about Progeria [NH](#) (National Human Genome Research Institute)
- Noonan Syndrome (Mayo Foundation for Medical Education and Research)
- Progeria 101/FAQ (Progeria Research Foundation, Inc.)
- Specific Genetic Disorders [NH](#) (National Human Genome Research Institute)
- Triple X Syndrome (Mayo Foundation for Medical Education and Research)
- Williams Syndrome [NH](#) (National Institute of Neurological Disorders and Stroke)

Genetics

- Genetics Home Reference: Genetic Conditions [NH](#) (National Library of Medicine)

Statistics and Research

Cystic Fibrosis
Down Syndrome
Dwarfism
Fragile X Syndrome
Genetic Brain Disorders
Genetic Counseling
Genetic Testing
Hemochromatosis
Leukodystrophies
Osteogenesis Imperfecta
Prader-Willi Syndrome
Rare Diseases
Sickle Cell Anemia

National Institutes of Health

The primary NIH organization for research on *Genetic Disorders* is the [National Institute of Child Health and Human Development](#)

NIH MedlinePlus Magazine

Medical Mysteries: "Thankful They Found a Cause"

Medical Mysteries: "We Feel Deep Compassion for Patients..."

Medical Mysteries: NIH Clinical Center. There's No Other Hospital Like It

Medical Mysteries: NIH Undiagnosed Diseases Program

Promise and Payoff of Rare Diseases Research

MedlinePlus



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Refine by Type

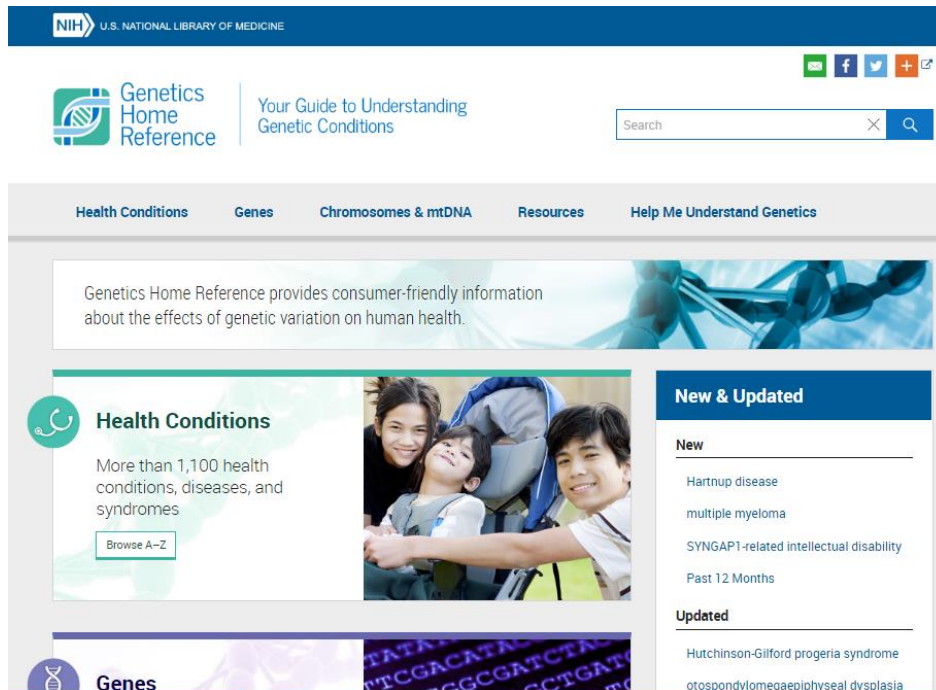
All Results (945)

- ☐ [Health Topics](#) (3)
- ☐ [External Health Links](#) (873)
- ☐ [Drugs and Supplements](#) (10)
- ☐ [Medical Encyclopedia](#) (11)
- ☐ [Videos and Tutorials](#)
- ☐ [News](#) (4)
- ☐ [MedlinePlus Magazine](#) (23)
- ☐ [Other Resources](#) (21)
- ☐ [Multiple Languages](#)

Results 1 - 10 of 945 for **genomics**


1. [Frequently Asked Questions about Genetic and **Genomic** Science](#)  (National Human Genome Research Institute)
... this page Frequently Asked Questions About Genetic and **Genomic** Science What are genetics and **genomics**? Why are ... **genomic** technologies? Additional Resources What are genetics and **genomics**? Genetics is a term that refers to the ...
<https://www.genome.gov/19016904> - External Health Links
2. [Genomics and Health Impact Update](#) (Centers for Disease Control and Prevention)
... Knowledge Base. What's New in the Public Health **Genomics** Knowledge Base New Implementation Tools Alzheimer's Disease Cardiomyopathy HIV/AIDS About the **Genomics** & Health Impact Update The Office of Public Health ...
www.cdc.gov/genomics/update/current.htm - External Health Links
3. [Brief Guide to **Genomics**: DNA, Genes and Genomes](#)  (National Human Genome Research Institute)
... Breve guía de genómica A Brief Guide to **Genomics** DNA, Genes and Genomes Deoxyribonucleic acid (DNA) is ... genetic basis for health and disease. Implications of **Genomics** for Medical Science Virtually every human ailment has ...
<https://www.genome.gov/18016863> - External Health Links
4. [Genomic Testing](#) (Centers for Disease Control and Prevention)

Genetics Home Reference




- Health conditions
- Genes
- Chromosomes and DNA
- Resources
- Genetic handbook

Genetics Home Reference



Health ConditionsGenesChromosomes & mtDNAResourcesHelp Me Understand Genetics



Health Conditions

Explore the signs and symptoms, frequency, genetic cause, and inheritance pattern of various conditions, diseases, and syndromes.

Browse by Category

Browse by First Letter

0-9

A

B

C

D

E

F

G

H

I

J

K

L

M

N

O

P

Q

R

S

T

U

V

W

X

Y

Z

A-[alphalipoprotein Neuropathy](#), see [Tangier disease](#)
A-T, see [ataxia-telangiectasia](#)
AAA, see [triple A syndrome](#)
AAA syndrome, see [triple A syndrome](#)
AADC deficiency, see [aromatic L-amino acid decarboxylase deficiency](#)
Aarskog syndrome, see [Aarskog-Scott syndrome](#)
[Aarskog-Scott syndrome](#)
AAS, see [Aarskog-Scott syndrome](#)
AASA dehydrogenase deficiency, see [pyridoxine-dependent epilepsy](#)
Aase syndrome, see [Diamond-Blackfan anemia](#)
Aase-Smith syndrome II, see [Diamond-Blackfan anemia](#)
AAT, see [alpha-1 antitrypsin deficiency](#)

Learn More about Health Conditions


What does it mean if a disorder seems to run in my family?

What are the different ways in which a genetic condition can be inherited?

What are complex or multifactorial disorders?

What does it mean to have a genetic predisposition to a disease?


Genetics Home Reference

Health ConditionsGenesChromosomes & mtDNAResourcesHelp Me Understand Genetics

breast cancer

Print AllOpen AllClose All

- Description
- Frequency
- Genetic Changes
- Inheritance Pattern
- Diagnosis & Management
- Other Names for This Condition
- Additional Information & Resources
- Sources for This Page
- Images

Health ConditionsGenesChromosomes & mtDNAResourcesHelp Me Understand Genetics

breast cancer

Print AllOpen AllClose All

- Description
- Frequency
- Genetic Changes
- ▼ Inheritance Pattern

Most cases of breast cancer are not caused by inherited genetic factors. These cancers are associated with somatic mutations in breast cells that are acquired during a person's lifetime, and they do not cluster in families.

In hereditary breast cancer, the way that cancer risk is inherited depends on the gene involved. For example, mutations in the *BRCA1* and *BRCA2* genes are inherited in an **autosomal dominant pattern**, which means one copy of the altered gene in each cell is sufficient to increase a person's chance of developing cancer. Although breast cancer is more common in women than in men, the mutated gene can be inherited from either the mother or the father.

In the other syndromes discussed above, the gene mutations that increase cancer risk also have an autosomal dominant pattern of inheritance. It is important to note that people inherit an increased likelihood of developing cancer, not the disease itself. Not all people who inherit mutations in these genes will ultimately develop cancer.

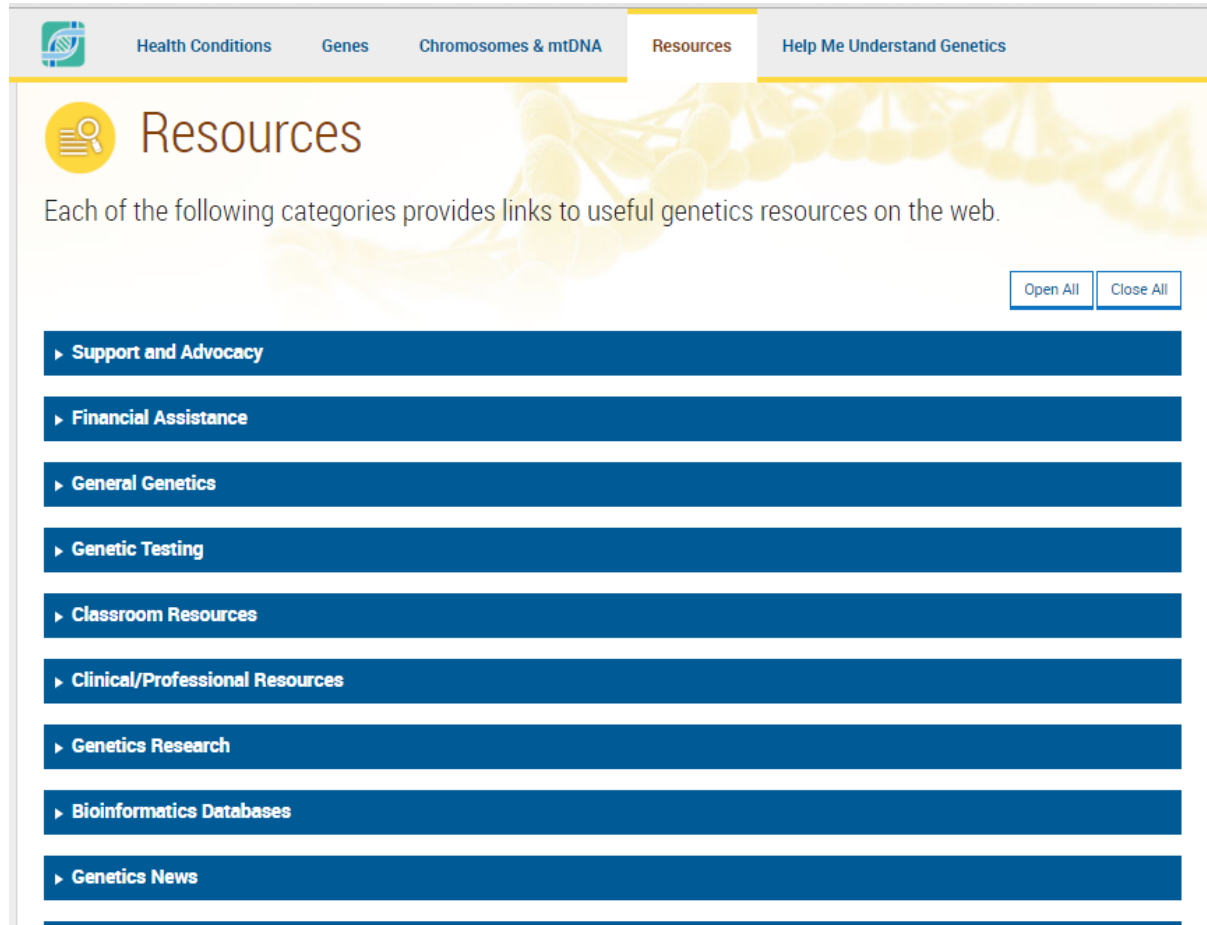
In many cases of breast cancer that clusters in families, the genetic basis for the disease and the mechanism of inheritance are unclear.

Related Information

- [What does it mean if a disorder seems to run in my family?](#)
- [What are the different ways in which a genetic condition can be inherited?](#)
- More about [Inheriting Genetic Conditions](#)

- Diagnosis & Management

Genetics Home Reference



The screenshot shows the 'Resources' page of the Genetics Home Reference website. The page has a navigation bar at the top with links to 'Health Conditions', 'Genes', 'Chromosomes & mtDNA', 'Resources' (which is highlighted), and 'Help Me Understand Genetics'. Below the navigation bar, the 'Resources' section is titled with a magnifying glass icon and the word 'Resources'. A paragraph states: 'Each of the following categories provides links to useful genetics resources on the web.' To the right of this paragraph are two buttons: 'Open All' and 'Close All'. Below this text is a list of ten resource categories, each in a blue bar with a right-pointing arrow: 'Support and Advocacy', 'Financial Assistance', 'General Genetics', 'Genetic Testing', 'Classroom Resources', 'Clinical/Professional Resources', 'Genetics Research', 'Bioinformatics Databases', and 'Genetics News'.

Health Conditions Genes Chromosomes & mtDNA **Resources** Help Me Understand Genetics


Resources


Each of the following categories provides links to useful genetics resources on the web.

[Open All](#) [Close All](#)

- ▶ Support and Advocacy
- ▶ Financial Assistance
- ▶ General Genetics
- ▶ Genetic Testing
- ▶ Classroom Resources
- ▶ Clinical/Professional Resources
- ▶ Genetics Research
- ▶ Bioinformatics Databases
- ▶ Genetics News

Genetics Home Reference

 [Health Conditions](#) [Genes](#) [Chromosomes & mtDNA](#) [Resources](#) [Help Me Understand Genetics](#)



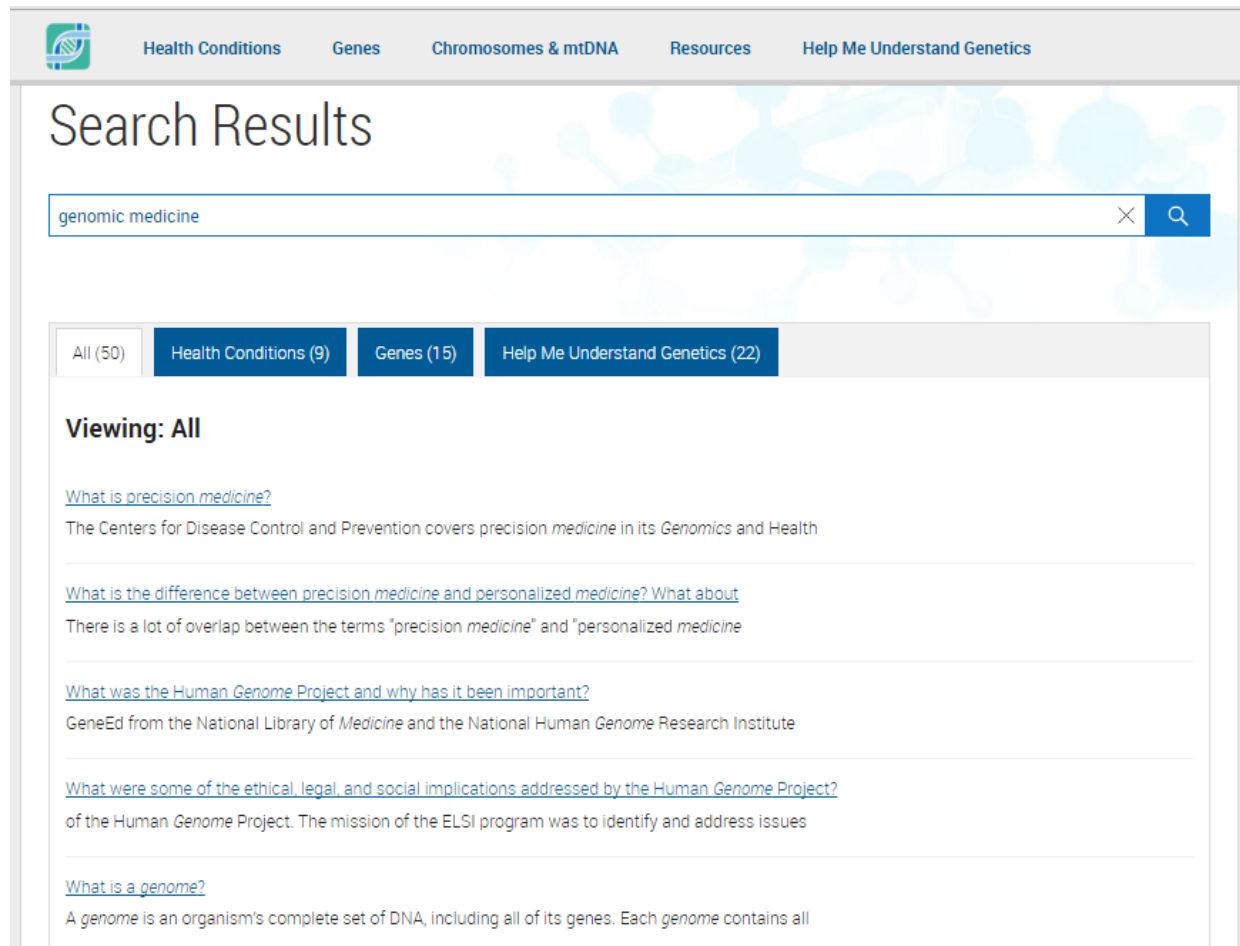
Help Me Understand Genetics

Help Me Understand Genetics provides an introduction to fundamental topics related to human genetics, including illustrations and basic explanations of genetics concepts.

[Download Book \(6MB\)](#) [Open All](#) [Close All](#)

- ▶ **Cells and DNA**
- ▶ **Mutations and Health**
- ▶ **How Genes Work**
- ▶ **Gene Families**
- ▶ **Inheriting Genetic Conditions**
- ▶ **Genetics and Human Traits**
- ▶ **Genetic Consultation**
- ▶ **Genetic Testing**

Genetics Home Reference



The screenshot shows the Genetics Home Reference website. At the top is a navigation bar with a logo and links for Health Conditions, Genes, Chromosomes & mtDNA, Resources, and Help Me Understand Genetics. Below this is a search bar containing the text 'genomic medicine'. The search results are displayed under the heading 'Search Results'. A filter bar shows 'All (50)' selected, with other options for 'Health Conditions (9)', 'Genes (15)', and 'Help Me Understand Genetics (22)'. The results are listed under the heading 'Viewing: All'. The first result is titled 'What is precision medicine?' and describes the Centers for Disease Control and Prevention's coverage of precision medicine. The second result is titled 'What is the difference between precision medicine and personalized medicine? What about' and discusses the overlap between the terms. The third result is titled 'What was the Human Genome Project and why has it been important?' and mentions GeneEd from the National Library of Medicine and the National Human Genome Research Institute. The fourth result is titled 'What were some of the ethical, legal, and social implications addressed by the Human Genome Project?' and discusses the mission of the ELSI program. The fifth result is titled 'What is a genome?' and defines a genome as an organism's complete set of DNA.

Health Conditions Genes Chromosomes & mtDNA Resources Help Me Understand Genetics

Search Results

genomic medicine

All (50) Health Conditions (9) Genes (15) Help Me Understand Genetics (22)

Viewing: All

[What is precision medicine?](#)
The Centers for Disease Control and Prevention covers precision *medicine* in its *Genomics* and Health





[What is the difference between precision medicine and personalized medicine? What about](#)
There is a lot of overlap between the terms "precision *medicine*" and "personalized *medicine*"

[What was the Human Genome Project and why has it been important?](#)
GeneEd from the National Library of *Medicine* and the National Human *Genome* Research Institute

[What were some of the ethical, legal, and social implications addressed by the Human Genome Project?](#)
of the Human *Genome* Project. The mission of the ELSI program was to identify and address issues


[What is a genome?](#)
A *genome* is an organism's complete set of DNA, including all of its genes. Each *genome* contains all

NIH National Human Genome Research Institute

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Health

Information about genetics and genomics, rare diseases, patient care and more



For Patients and the Public


- [Community Engagement and Community Health](#)
- [Family History](#)
- [Genetics & Genomics Science & Research](#)
- [Genetic & Rare Diseases Information Center](#)
- [Genomic Medicine and Health Care](#)
- [Online Health and Support Resources](#)
- [Specific Genetic Disorders](#)

For Health Professionals

- [Competency & Curricular Resources](#)
- [Genetics 101](#)
- [Inter-Society Coordinating Committee \(ISCC\)](#)
- [New Horizons and Research](#)
- [Patient Management](#)
- [Policy and Ethics Issues](#)
- [What is Genomic Medicine?](#)

Feature

Apply now for the "short course" health professionals track




The Genomic Healthcare Branch is offering the NHGRI Short Course in Genomics: Nurse, Physician Assistant and Faculty Track from August 1 - August 3, 2016. This year's course is for nurses, nurse practitioners, physician assistants and the faculty who educate these health professionals in genomics. [Read more](#)

Webinars for Health Insurers and Payers: Understanding Genetic Testing

Highlights

Video: *A G2C2 Website Overview*



This introduction to the Genetics/Genomics Competency Center (G2C2) website at <http://g-2-c-2.org/>, gives new users an opportunity to view key features of this centralized collection of genomics educational resources for healthcare educators and providers. [Read more](#)


Genomic knowledge is power in the fight against obesity

Although many doctors are

See Also

- [GenomeTV](#)
- [Genomic Healthcare Branch](#)
- [Fact Sheets](#)
- [Genetic Education Resources for Teachers](#)
- [All About the Human Genome Project](#)
- [Health Archive](#)

On Other Sites:

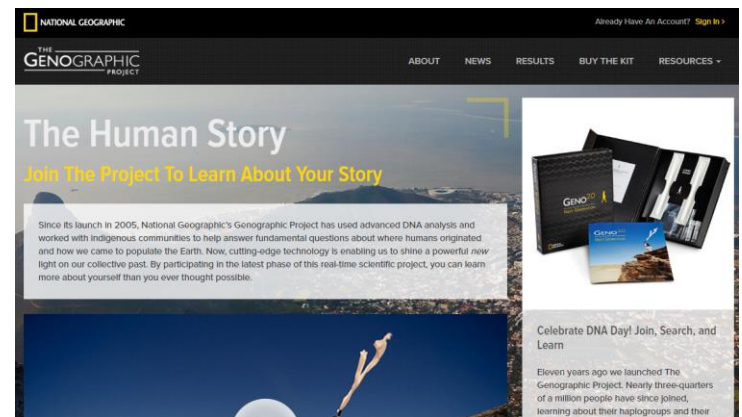
 [GenomeTV](#)
NHGRI's YouTube channel



Direct to Consumer Testing

Genetic Testing

Direct to Consumer Testing



Concerns

- Privacy
- Legality
- Who has access?
- How useful now?
- What all is being done now and in the future with the information?
- Unexpected surprises?
- Test results can vary among companies
- Validity of tests
- No counseling provided
- Who can get the testing?

Benefits



- ❑ Learn more about own health
- ❑ More effective medical treatments
- ❑ Learn more about ethnicity and family history
- ❑ Bring awareness to family health issues for future generations
- ❑ Motivation to work on health habits
- ❑ Encourages patient engagement
- ❑ Contributing to advancement of healthcare and science
- ❑ Moral obligation



Example of a 23andMe test result

Genetics Home Reference

NIH U.S. NATIONAL LIBRARY OF MEDICINE



Your Guide to Understanding Genetic Conditions



Health Conditions

Genes

Chromosomes & mtDNA

Resources

Help Me Understand Genetics

Home

Help Me Understand Genetics

Genetic Testing


What is direct-to-consumer genetic testing?

What is direct-to-consumer genetic testing?

Traditionally, genetic tests have been available only through healthcare providers such as physicians, nurse practitioners, and genetic counselors. Healthcare providers order the appropriate test from a laboratory, collect and send the samples, and interpret the test results. Direct-to-consumer genetic testing refers to genetic tests that are marketed directly to consumers via television, print advertisements, or the Internet. This form of testing, which is also known as at-home genetic testing, provides access to a person's genetic information without necessarily involving a doctor or insurance company in the process.


If a consumer chooses to purchase a genetic test directly, the test kit is mailed to the consumer instead of being ordered through a doctor's office. The test typically involves collecting a DNA sample at home, often by swabbing the inside of the cheek, and mailing the sample back to the laboratory. In some cases, the person must visit a health clinic to have blood drawn. Consumers are notified of their results by mail or over the telephone, or the results are posted online. In some cases, a genetic counselor or other healthcare provider is available to explain the results and answer questions. The price for this type of at-home genetic testing ranges from several hundred dollars to more than a thousand dollars.

For more information about direct-to-consumer genetic testing:

The American College of Medical Genetics, which is a national association of doctors specializing in genetics, has issued [a statement on direct-to-consumer genetic testing](#) .

The American Society of Human Genetics, a professional membership organization for specialists in genetics, has also issued [a statement on direct-to-consumer genetic testing in the United](#)

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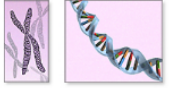
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Home → [Health Topics](#) → [Genetic Testing](#)

Genetic Testing

On this page

Basics <ul style="list-style-type: none">SummaryStart HereLatest News	Learn More <ul style="list-style-type: none">Related IssuesSpecifics	See, Play and Learn <ul style="list-style-type: none">No links available
Research <ul style="list-style-type: none">Statistics and ResearchClinical TrialsJournal Articles	Resources <ul style="list-style-type: none">Reference DeskFind an Expert	For You <ul style="list-style-type: none">Patient Handouts

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MEDICAL ENCYCLOPEDIA

- [BRCA1 and BRCA2 gene testing](#)
- [Buccal smear](#)
- [Genetic testing and your cancer risk](#)

Related Health Topics

- [Birth Defects](#)

Summary

Genetic tests are tests on blood and other tissue to find [genetic disorders](#). Over 2000 tests are available. Doctors use genetic tests for several reasons. These include

- Finding genetic diseases in unborn babies
- Finding out if people carry a gene for a disease and might pass it on to their children
- Screening embryos for disease
- Testing for genetic diseases in adults before they cause symptoms
- Making a diagnosis in a person who has disease symptoms
- Figuring out the type or dose of a medicine that is best for a certain person

People have many different reasons for being tested or not being tested. For some, it is important to know whether a disease can be prevented or treated if a test is positive. In some cases, there is no treatment. But test results might help a person make life decisions, such as family planning or insurance coverage. A [genetic counselor](#) can provide information about the pros and cons of testing.

NIH: National Human Genome Research Institute

insurance coverage. A genetic counselor can provide information about the pros and cons of testing.

NIH: National Human Genome Research Institute

Start Here

- [Frequently Asked Questions about Genetic Testing](#) (NIH) (National Human Genome Research Institute)
Available in Spanish
- [Genetic Testing \(For Parents\)](#) (Nemours Foundation)
Available in Spanish
- [Genetic Testing: What You Should Know](#) (American Academy of Family Physicians)
Available in Spanish
- [Regulation of Genetic Tests](#) (NIH) (National Human Genome Research Institute)
Available in Spanish

Latest News

- [Genetic Insights May Help Kids Battling Developmental Delays](#) (05/25/2016, HealthDay)
- [Researchers Find 8 Immune Genes in Aggressive Brain Cancer](#) (05/25/2016, HealthDay)
- [Two Genes May Raise Odds for Fraternal Twin Pregnancies](#) (04/28/2016, HealthDay)
- [More News on Genetic Testing](#)

Related Issues

- [Direct-to-Consumer Genetic Tests](#) (Federal Trade Commission)
Available in Spanish
- [Genetics Home Reference](#) (NIH) (National Library of Medicine)
- [Genomic Testing](#) (Centers for Disease Control and Prevention)
- [How Can Consumers Be Sure a Genetic Test Is Valid and Useful?](#) (NIH) (National Library of Medicine)
- [How Is Genetic Testing Done?](#) (NIH) (National Library of Medicine)
- [Personalized Medicine and Pharmacogenomics](#) (Mayo Foundation for Medical Education and Research)
- [Studying Genes](#) (NIH) (National Institute of General Medical Sciences)
- [What Are the Risks and Limitations of Genetic Testing?](#) (NIH) (National Library of Medicine)
- [What Do the Results of Genetic Tests Mean?](#) (NIH) (National Library of Medicine)
- [What Is a Pediatric Geneticist?](#) (American Academy of Pediatrics)
Available in Spanish
- [What Is Direct-to-Consumer Genetic Testing?](#) (NIH) (National Library of Medicine)
- [Whole Genome Sequencing](#) (Genetic Alliance)

Related Health Topics

- [Birth Defects](#)
- [Genetic Counseling](#)
- [Genetic Disorders](#)
- [Newborn Screening](#)
- [Prenatal Testing](#)

National Institutes of Health

The primary NIH organization for research on *Genetic Testing* is the [National Human Genome Research Institute](#)

NIH MedlinePlus Magazine

[Future of Personalized Medicine](#)

MedlinePlus links to health information from the National Institutes of Health and other federal government agencies. MedlinePlus also links to health information from non-government Web sites. See our [disclaimer](#) about external links and our quality guidelines.

American College of Medical Genetics and Genomics

Folic Acid

Folic Acid and Neural Tube Defects	2010, ACMG	2010 Genet Med 13:6:593-596
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Genetic Discrimination

Points to Consider in Preventing Unfair Discrimination Based on Genetic Disease Risk: A Position Statement of the American College of Medical Genetics and Genomics	2001, ACMG Reaffirmed 2005	2001 Genet Med 3:6: 436-437
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Genetic Services

Clinical utility of genetic and genomic services: a position statement of the American College of Medical Genetics and Genomics	2015, ACMG	2015 Genet Med 7:6:505-507
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Genetic Testing

ACMG Revised Position Statement on Direct-to-Consumer Genetic Testing	2015, ACMG	2016 Genet Med 18:2:207-208
ACMG position statement on prenatal/ preconception expanded carrier screening	2013, ACMG	2013 Genet Med 15:6:482-483
Technical report: ethical and policy issues in genetic testing and screening of children	2013, ACMG/AAP	2013 Genet Med 15:3:234-245
Risk categorization for oversight of laboratory-developed tests for inherited conditions	2013, ACMG	2013 Genet Med 15:4:314-5
Genetic Testing in Adoption (ACMG/ASHG)	2000, ASHG	2000 Am J Hum Genet 66:761-767

Genomic Sequencing

ACMG Policy Statement: Updated Recommendations Regarding Analysis and Reporting of Secondary Findings in Clinical Genome-Scale Sequencing	2015, ACMG	2015 Genet Med 17:1:68-69
ACMG Recommendations for reporting of incidental findings in clinical exome and	2013,	2013 Genet

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ACMG STATEMENT | Genetics in Medicine

Direct-to-consumer genetic testing: a revised position statement of the American College of Medical Genetics and Genomics

ACMG Board of Directors¹

Disclaimer: These recommendations are designed primarily as an educational resource for medical geneticists and other health-care providers to help them provide quality medical genetics services. Adherence to these recommendations does not necessarily assure a successful medical outcome. These recommendations should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, geneticists and other

clinicians should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from the recommendations.

Genet Med advance online publication 17 December 2015

Key Words: consumer; direct-to-consumer; genetic testing; self-testing; OTC

With ongoing genetic discoveries and improvements in technology, more genetic tests are available than ever before. Along with greater availability has come increased consumer demand for genetic tests and expansion of direct-to-consumer testing. The American College of Medical Genetics and Genomics (ACMG) has revised its 2008 e-publication regarding this issue (ACMG Statement on Direct-to-Consumer Genetic Testing, retired; available by request to acmg@acmg.net) and believes that it is critical for the public to realize that genetic testing is only one part of a complex process that includes genetic risk

A genetics expert such as a certified medical geneticist or genetic counselor should be available to help the consumer determine, for example, whether a genetic test should be performed and how to interpret test results in light of personal and family history. A board-certified genetic counselor can help facilitate this process by providing information about the test and helping to explain test results. A number of risks can be reduced if a board-certified genetics professional is involved in genetic testing, including inadequate or lack of informed consent

ACMG <https://www.acmg.net/>



Ethics and Privacy

Societal Concerns

- Who should have access to personal genetic information, and how will it be used?
- Who owns and controls genetic information?
- How does personal genetic information affect an individual and society's perceptions of that individual?
- What are the larger societal issues raised by new reproductive technologies?
- How will genetic tests be evaluated and regulated for accuracy, reliability and utility?
- How do we prepare healthcare professionals and the public?
- What is considered acceptable diversity?
- Where is the line between medical treatment and enhancement?
- Should testing be performed when no treatment is available?

GINA

GINA

GENETIC INFORMATION
NONDISCRIMINATION ACT

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Genetic Information

What is genetic information and why is it important?

GINA & Health Insurance

What are GINA's health insurance protections?

GINA & Employment

What are GINA's employment protections?

What is GINA?

The Genetic Information Nondiscrimination Act of 2008 (GINA) is a federal law that protects individuals from genetic discrimination in health insurance and employment. Genetic discrimination is the misuse of genetic information. This resource provides an introduction to GINA and its protections in health insurance and employment. It includes answers to common questions and examples to help you learn. Choose from one of the boxes to the left to begin!

✉ Have questions, comments or suggestions? [Send us a note.](#)

🖨 [Click here](#) for a printer friendly version.

✉ For healthcare provider resources [click here.](#)

🖨 [Click here](#) for the GINA & You Information Sheet

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Catalog item

Choosing Wisely

<http://consumerhealthchoices.org/catalog/making-smart-decisions-about-genetic-testing-acmg/>

Making Smart Decisions About Genetic Testing (ACMG)

Campaign Series: Choosing Wisely

Medical Category: Tests

Article Type: Advice

Language: Plain English, Spanish

Affiliation: American College of Medical Genetics and Genomics

Format: HTML, PDF

Most recent update: 10/07/2015



Sometimes a genetic test is not the best way to find an inherited condition or disease risk. A routine blood test or procedure might be just as good. And it might be less costly and more easily available.

Files to download

Plain English: [Making Smart Decisions About Genetic Testing \(ACMG\) →](#)

Spanish: [Cómo tomar decisiones inteligentes sobre las pruebas genéticas →](#)

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Webpage file: [Making Smart Decisions About Genetic Testing →](#)

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ACMG NEWS For Immediate Release

Contact: Kathy Beal, MBA
ACMG Media Relations
301-238-4582, kbeal@acmg.net

ACMG Provides Recommendations on Genetic Testing Through the Choosing Wisely® Campaign

Goal is to encourage clinician and patient conversations about appropriate genetic testing


Bethesda, MD -- July 10, 2015- The American College of Medical Genetics and Genomics (ACMG) has released a list of five things patients and providers should discuss regarding specific genetic tests as part of *Choosing Wisely*, an initiative of the ABIM Foundation. The just-released list identifies five evidence-based recommendations that can support conversations between patients and clinicians regarding genetic testing. To date, more than 100 national and state medical specialty societies, regional health collaboratives and consumer partners have joined the *Choosing Wisely* campaign about appropriate care.


The list provided by ACMG provides evidence-based recommendations to help all clinicians and patients to have conversations about making wise choices related to genetic testing. The lists were developed over the past year with careful consideration of the latest evidence, expert opinions and research.


ACMG





https://www.acmg.net/docs/ACMG_ChoosingWisely_Final.pdf

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 **National Human Genome Research Institute**
Advancing human health through genomics research


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Issues in Genetics | Policy, legal and ethical issues in genetic research





 Coverage and Reimbursement in Genetic Tests


 Regulation of Genetic Tests
Regulations, congressional activity, resources


 Intellectual Property and Genomics
Gene patenting, guidelines


 Human Subjects Research
Protections, return of results, more


 Privacy in Genomics
Privacy in research, the clinic, society, more


 Genetics and Public Policy Fellowship

 Genetic Discrimination
GINA, Statutes, Regulations, more

 Informed Consent
Process, sample language, consent forms, more

 Genome Statute and Legislation Database
Search federal & state law

Feature
Advisory committee addresses building a medical information commons

While generating large amounts of medical and genomic data may improve health, questions about access and use remain. To address these challenges, an

Highlights
Investigational Device Exemptions (IDE) in Genomic Research Workshop

On June 10, 2016, the National Human Genome Research Institute (NHGRI) will host a day-long, public workshop - *Investigational Device Exemptions (IDE) and Genomic Research*. The purpose of this workshop is educational. The Food and Drug Administration (FDA) may require

See Also
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NHGRI Extramural Research Program

[GenomeTV](#)

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Past Web content from the Issues in Genetics Portal

American Academy of Pediatrics American College of Medical Genetics and Genomics

Pediatrics

March 2013, VOLUME 131 / ISSUE 3

From the American Academy of Pediatrics

Policy Statement

AAP

<http://pediatrics.aappublications.org/content/131/3/620>

Ethical and Policy Issues in Genetic Testing and Screening of Children

COMMITTEE ON BIOETHICS, COMMITTEE ON GENETICS, AND, THE AMERICAN COLLEGE OF MEDICAL GENETICS AND, GENOMICS SOCIAL, ETHICAL, AND LEGAL ISSUES COMMITTEE

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Abstract

The genetic testing and genetic screening of children are commonplace. Decisions about whether to offer genetic testing and screening should be driven by the best interest of the child. The growing literature on the psychosocial and clinical effects of such testing and screening can help inform best practices. This policy statement represents recommendations developed collaboratively by the American Academy of Pediatrics and the American College of Medical Genetics and Genomics with respect to many of the scenarios in which genetic testing and screening can occur.

ACMG

https://www.acmg.net/docs/genetic_testing_in_children_preprint_gim2012176a.pdf

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ACMG POLICY STATEMENT

Genetics
inMedicine

Technical report: ethical and policy issues in genetic testing and screening of children

Laine Friedman Ross, MD, PhD¹⁻², Howard M. Saal, MD³, Karen L. David, MD, MS^{4,6} and Rebecca R. Anderson, JD, MS⁵; and the American Academy of Pediatrics; American College of Medical Genetics and Genomics

The genetic testing and genetic screening of children are commonplace. Decisions about whether to offer genetic testing and screening should be driven by the best interest of the child. The growing literature on the psychosocial and clinical effects of such testing and screening can help inform best practices. This technical report provides ethical justification and empirical data in support of the pro-

posed policy recommendations regarding such practices in a myriad of settings.

Genet Med advance online publication 21 February 2013

Key Words: carrier identification; disclosure; genetic screening; genetic testing; newborn screening; predictive testing

INTRODUCTION

Two major events occurred in the 1950s that forever changed the influence of genetics in medicine: Watson and Crick¹ described the double-helix model of DNA structure in 1953, and in 1956 Tjio and Levan² established that the typical human carries 46 chromosomes. The goal of mapping and sequencing the human genome began in 1990, and a working draft was presented in 2000, with a more complete edition published in 2003.³ Knowledge of genetics and genomics continues to grow rapidly, as does consumer interest in genetic testing. As a result, statements about genetic testing and screening of children in the United States written in the past two decades need to be updated to consider the ethical issues that arise with the new technologies and expanded uses of genetic testing and screen-

ing, and endocrine abnormalities for which early treatment may prevent or reduce morbidity or mortality. Most of the genetic conditions included in the state screening panels are autosomal recessive disorders, and some assays identify heterozygote carriers (e.g., hemoglobinopathies). Future screening may expand to X-linked conditions (e.g., Duchenne muscular dystrophy) and autosomal dominant conditions. In addition, universal newborn hearing screening allows for early identification of both acquired and hereditary hearing loss.

Outside of newborn screening, pediatric genetic testing is much less common. Diagnostic genetic testing may be performed on a child with physical, developmental, or behavioral features consistent with a potential genetic syndrome or for pharmacogenetic drug selection and dosing decisions.

Informing the Public



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GENETICS AND
SOCIETY



Precision Medicine

“...a bold new research effort to revolutionize how we improve health and treat disease.”

[PMI announcement](https://www.whitehouse.gov/precision-medicine) <https://www.whitehouse.gov/precision-medicine>

Precision Medicine Initiative

Mission statement:

To enable a new era of medicine through research, technology, and policies that empower patients, researchers, and providers to work together toward development of individualized care.



THE PRECISION MEDICINE INITIATIVE



PRECISION MEDICINE INITIATIVE PRINCIPLES STORIES

f t GO TO TOP

[PMI announcement](https://www.whitehouse.gov/precision-medicine) <https://www.whitehouse.gov/precision-medicine>

Precision Medicine is...



- Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.
- Instead of what treatment is right for this disease it is what treatment is right for this patient.

Precision Medicine Initiative

□ **Near Term goals:**

- Clinical trials focusing on pediatric cancers and drug therapies for adults
- Use of combination therapies
- Overcoming drug resistance

□ **Long Term Goals:**

- Create research cohort of 1 million volunteers
- New model of medicine
 - engage participants
 - responsible data sharing
 - privacy protection
- Advance pharmacogenomics
- Identify new targets for treatment and prevention
- Test if mobile devices encourages healthy behaviors
- Lay scientific foundation for many diseases

Precision Medicine Cohort Program

- Two ways to participate
 1. Through the cohort website
 2. With participating health care provider organization



**PRECISION MEDICINE INITIATIVE®
COHORT PROGRAM**

WHAT IS IT?

Precision medicine is a groundbreaking approach to disease prevention and treatment based on people's individual differences in environment, genes and lifestyle.

The Precision Medicine Initiative® Cohort Program will lay the foundation for using this approach in **clinical practice**.

WHAT ARE THE GOALS?

Engage a group of **1 million or more U.S. research participants** who will share biological samples, genetic data and lifestyle information, all linked to their electronic health records. This data will allow researchers to develop more precise treatments for **many diseases and conditions**.

Pioneer a new model of research that emphasizes **engaged research participants, responsible data sharing and privacy protection**.

Research based on the cohort data will:

- Lay **scientific foundation** for precision medicine
- Help identify new ways to **treat and prevent disease**
- Test whether **mobile devices**, such as phones and tablets, can encourage healthy behaviors
- Help develop the **right drug** for the **right person** at the **right dose**

WHY NOW?

The **time is right** because:

We have a greater understanding of human genes	People are more engaged in healthcare and research
We have the tools to track health information and use large databases	Research technologies have improved

Follow the Initiative's progress and be one of the first to join this landmark effort.

NIH and Precision Medicine Initiative

PRECISION MEDICINE INITIATIVE COHORT PROGRAM

Precision Medicine Initiative

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NIH awards \$55 million to build million-person precision medicine study



Learn about the key components of the PMI Cohort Program

About the Precision Medicine Initiative Cohort Program

Far too many diseases do not have a proven means of prevention or effective treatments. We must gain better insights into the biological, environmental, and behavioral influences on these diseases to make a difference for the millions of Americans who suffer from them. Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person. While some advances in precision medicine have been made, the practice is not currently in use for most diseases.



Email Updates

Sign up to receive email updates about the Precision Medicine Initiative.

[Sign up for updates](#)

Related Links

[PMI Working Group Final Report](#)

[pdf](#)

[NEJM Perspective: A New Initiative on Precision Medicine](#)

[White House Precision Medicine Web Page](#)

[White House Fact Sheet: President Obama's Precision Medicine Initiative](#)

[Precision Medicine Initiative and Cancer Research](#)

[Precision Medicine Initiative YouTube Channel](#)

[PMI Cohort Program ://www.nih.gov/precision-medicine-initiative-cohort-program](http://www.nih.gov/precision-medicine-initiative-cohort-program)

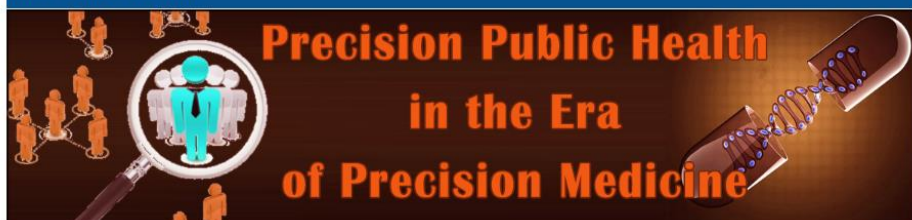
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Health Care Tailored to You



Public Health Genomics



What's New in Public Health Genomics Knowledge Base



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BIG DATA



HEPATITIS



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WEEKLY UPDATE

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Genomics and Health Impact Blog Posts



The Shift From Personalized Medicine to Precision Medicine and Precision Public Health: Words Matter!



Advances in genomics and other 'omic' technologies have ushered in a new era variably called "personalized" or "precision" medicine, which takes into account individual genetic and other sources of variability in disease treatment and prevention. In the past decade, we have seen a significant growth in interest and usage of the terms personalized and precision [Read More >](#)

Posted on April 21, 2016 by Muin J Khoury, Director, Office of Public Health Genomics, Centers for Disease Control and Prevention

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Does genetic risk information improve healthy behavior? Let's not throw out the baby with the bath water!

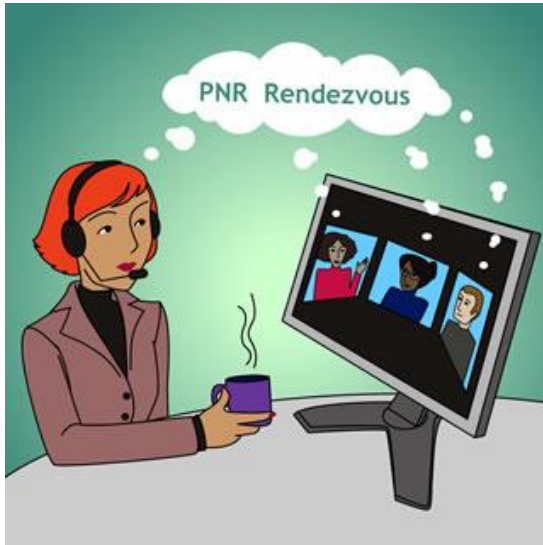


In a recent systematic review with meta-analysis, Hollands et al evaluated the impact of communicating genetic risk information on risk-reducing health behaviors and motivations for behavior change. The authors reviewed 18 studies with 7

[Genomics health blog](http://blogs.cdc.gov/genomics/)

<http://blogs.cdc.gov/genomics/>

PNR Rendezvous



Informed Consent and the Precision Medicine Initiative September 21, 2016 1PM

Presenter: Malia Fullerton,
Associate Professor of Bioethics and
Humanities at the University of
Washington School of Medicine

Library role

- “Preparing the public to make educated personal and family health decisions in a time of rapidly evolving genetic and genomic knowledge will require new partnerships between the education system, health care systems, the government, community advocacy organizations, consumers and the media.”

Show What You Know!

- The 1000 Genomes Project was undertaken in order to increase the _____ of the genomes represented in public databases.
- What term refers to strategies for determining what treatment is right for an INDIVIDUAL rather than what treatment is recommended for a DISEASE?
- Clinicians are not concerned about all genetic variants – only those that are _____.
- True or False? GINA (Genetic Information Nondiscrimination Act) protects you from life insurance discrimination.
- True or False? A genetic variant may originally be classified as “likely pathogenic” and later classified as “likely benign.”
- What resource would you recommend to consumers who wanted to learn more about a genetic condition?
- What is a good starting place for finding genetic information for clinicians?

Questions?

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Presentation resources

<https://nnlm.gov/pnr/training/presentations>

